

**CLAIMS**

1. A method of associating a genotype with a phenotype, comprising:
  - determining levels of expression of an allele of a gene in a first population comprising affected individuals, said affected individuals sharing a phenotype;
  - determining levels of expression of the allele in a second population comprising control individuals, said control individuals not sharing the phenotype;
  - comparing levels of expression of the allele in the first and the second populations;
  - identifying an allele whose expression differs in a statistically significant manner between the first and the second populations as having an association with the phenotype.
2. The method of claim 1 wherein the phenotype is a disease susceptibility.
3. The method of claim 1 wherein the phenotype is a disease.
4. The method of claim 1 wherein the phenotype is a birth defect.
5. The method of claim 1 wherein the affected individuals are heterozygous for the gene.
6. The method of claim 1 wherein the control individuals are heterozygous for the gene.
7. The method of claim 1 wherein the phenotype is a polymorphic phenotype.
8. The method of claim 1 wherein expression of the allele is determined independent of the expression of other alleles of the gene.
9. The method of claim 1 wherein the phenotype is not related to a known disease.
10. The method of claim 1 further comprising determining a haplotype associated with the allele in the first population.
11. The method of claim 1 wherein the level of expression of the allele is heritable.
12. The method of claim 1 further comprising determining a sequence variation which is associated with the allele in the first population.
13. The method of claim 12 wherein the sequence variation is a single nucleotide polymorphism (SNP).
14. The method of claim 12 wherein the sequence variation is an insertion.
15. The method of claim 12 wherein the sequence variation is a deletion.

16. The method of claim 12 further comprising determining that the sequence variation causes the level of expression of the allele to differ from level of expression of at least one other allele of the gene.
17. The method of claim 1 wherein the levels of expression are determined and compared using fluorescent dye terminators and a single-base extension reaction.
18. The method of claim 17 wherein the levels of expression are determined and compared using capillary electrophoresis.
19. A method of measuring allelic expression variation in a non-imprinted gene in a first individual, comprising:
  - reverse transcribing and amplifying* mRNA from an individual heterozygous for a single nucleotide polymorphism (SNP) in a non-imprinted gene to form first cDNA from a first allele and second cDNA from a second allele;
  - hybridizing* primers to first cDNA and second cDNA *and differentially labeling* those primers hybridized to first cDNA and second cDNA to form differentially labeled first and second primers;
  - comparing* amount of differentially labeled first primers to amount of differentially labeled second primers, wherein a statistically significant difference in the amount of labeled first primers from the amount of labeled second primers indicates that the first and second alleles are differentially expressed in the first individual.
20. The method of claim 19 wherein the differential labeling is performed using fluorescent dye terminators.
21. The method of claim 19 wherein the comparing is performed using capillary electrophoresis.
22. The method of claim 19 wherein the differential labeling is performed using a single base extension reaction.
23. The method of claim 19 further comprising measuring allelic expression of the first or second allele in a second individual related to the first individual to confirm that the allelic expression variation is heritable.
24. The method of claim 23 wherein the second individual is a parent or offspring of the first individual.

25. The method of claim 23 wherein the first and second alleles are both expressed in the first individual.
26. The method of claim 19 wherein the statistically significant difference is at least 20%.
27. The method of claim 19 further comprising determining a haplotype associated with the first allele in the first individual.
28. The method of claim 19 further comprising determining a sequence variation which is associated with the first allele in the first individual.
29. The method of claim 28 wherein the sequence variation is a single nucleotide polymorphism (SNP).
30. The method of claim 28 wherein the sequence variation is an insertion.
31. The method of claim 28 wherein the sequence variation is a deletion.
32. A method of measuring allelic expression variation in a non-imprinted gene in a first individual, comprising:
  - reverse transcribing and amplifying* mRNA from an individual heterozygous for a single nucleotide polymorphism (SNP) in a non-imprinted gene to form first cDNA from a first allele and second cDNA from a second allele;
  - hybridizing* primers to first cDNA and second cDNA *and differentially labeling* those primers hybridized to first cDNA and second cDNA using fluorescent dye terminators and a single base extension reaction to form differentially labeled first and second primers;
33. *comparing* amount of differentially labeled first primers to amount of differentially labeled second primers using capillary electrophoresis, wherein a statistically significant difference in the amount of labeled first primers from the amount of labeled second primers indicates that the first and second alleles are differentially expressed in the first individual.
34. A method of measuring allelic expression variation in a non-imprinted gene in a first individual, comprising:
  - determining level of expression of an allele of a gene in a first individual displaying a phenotype;
  - determining level of expression of the allele in a population of control individuals, said control individuals not displaying the phenotype;

comparing level of expression of the allele in the first individual to level of expression in the population of control individuals, wherein a statistically significant difference in the levels of expression indicates that the allele in the first individual may be associated with the phenotype.

35. A method of measuring allelic expression variation in a non-imprinted gene in a first individual, comprising:

determining level of expression of an allele of a gene in a first individual, wherein a level of expression of the gene has been associated with a phenotype;

comparing level of expression of the allele in the first individual to level of expression in a first or second population of control individuals, wherein the first population of control individuals have the phenotype and wherein the second population of control individuals do not have the phenotype, wherein a statistically significant difference in the levels of expression between the first individual and the second population indicates that the first individual has the phenotype and wherein no statistically significant difference in the levels of expression between the first individual and the first population indicates that the first individual does not have the phenotype.